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184 Evaluation of the impact of SERPINA gene mutation on the occurrence of liver damage and cholestasis in patients diagnosed with cystic fibrosis

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SERPINA1 gene is present in about 2% of patients with cystic fibrosis, but more frequently, in approximately 5% of patients with cystic fibrosis (CF) and concomitant changes in the liver.

Aim: The aim of this study was to assess the impact of SERPINA gene mutation on the occurrence of liver damage and cholestasis in patients diagnosed with cystic fibrosis.

Patients and Methods: The analysis included 30 children, 13 girls (43.3%) and 17 boys (56.6%), aged from 6 months to 18 years, with diagnosed CF. All patients underwent genetic testing for SERPINA gene mutation. The analysis included age, sex, clinical symptoms, type of mutation of the CFTR protein, laboratory abnormalities (levels of transaminases, GGTP, FA, protein, *acid steatocrit*) and ultrasound examination of the abdomen.

Results: Elevated transaminases were found in 9/30 (30%), whereas elevated levels of gamma-glutamyl transferase in 6/30 (20%) children. In 5/30 patients the ultrasound examination demonstrated liver enlargement with increased echogenicity. The mutation in SERPINA gene was found in 1/30 (3.3%) patient with cystic fibrosis. Currently, this patient has normal values of transaminases, GGTP and FA, whereas, a significant worsening of respiratory symptoms is observed. There was no correlation between the occurrence of SERPINA gene mutation and clinical symptoms, type of CFTR protein mutation, results of laboratory tests of liver function and hepatocyte damage, and ultrasound examination of the abdomen.

Conclusions: There was no correlation between the occurrence of SERPINA gene mutation and the presence of features of liver damage and cholestasis in children diagnosed with cystic fibrosis.

185 Transient elastography measurements in adults with cystic fibrosis liver disease

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Objectives: Liver disease causes an increase in its stiffness and although this can be measured by transient elastography (TE), the utility of this simple test in adult CF-related liver disease (CFLD) has not been established. We report the use of TE to assess CFLD in the largest series of adult CF patients to date.

Methods: Using a Fibroscan® 402, TE was measured by taking the median value from at least 10 readings. The diagnosis of CFLD in individual patients was confirmed from blood, ultrasound and endoscopy results.

Results: Of 139 consecutive patients studied (mean age 27.3 [range 17–55], 85 male), valid readings were obtained in 136 (98%) but 4 patients had incomplete datasets. TE (in kPa) of the 22 with CFLD was higher than the remainder (median 11.1 [IQR 8.3–13.7] versus 4.3 [3.5–5.4], $p < 0.001$). TE was a highly accurate test for predicting CFLD (area under receiver operating curve = 0.95): a value <6.6 kPa effectively excluded the diagnosis (99% negative predictive value, negative likelihood ratio 0.05) whilst those >7.6 kPa were at least 30 times more likely to have liver disease.

	Mean age	% Male	Mean FEV1 %	CFRD	Median TE (IQR)
CFLD (n = 22)	26.0	77.3%	64.5%	50.0%	11.1 kPa (8.3–13.7)
No CFLD (n = 110)	27.5	59.1%	65.9%	68.2%	4.3 kPa (3.5–5.4)

Conclusion: In this, the largest series of its kind, we have shown that TE, a simple non-invasive test, can be performed during routine outpatient review, and is a highly accurate predictor of CFLD in adult CF patients.

186 Transient elastography can be used to guide monitoring of cystic fibrosis related liver disease in adult patients

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Objectives: The optimal frequency of ultrasound scans (USS) to monitor the development of portal hypertension (PH) in CF-related liver disease (CFLD) is unknown. Transient elastography (TE), a simple non-invasive form of monitoring, is being increasingly used as an alternative to USS but there is little experience in adult CF.

Methods: Using a Fibroscan® 402 device, we compared the utility of TE with USS in the diagnosis of PH. TE was measured using the median value from at least 10 readings at routine outpatient review. The most recent USS was reviewed and considered abnormal if it featured heterogeneous or nodular changes or signs of portal hypertension.

Results: TE was compared with contemporaneous USS in 78 patients (mean age 26.8, [range 17–53], male 66.7%). The 15 with an abnormal USS had a higher TE than the remainder (median 11.7 kPa [IQR 10.0–19.6] vs 4.4 [IQR 3.7–6.3] $p < 0.005$). TE was highly useful in differentiating whether abnormalities were present on USS (area under receiver operating curve = 0.94), where a cut-off of 6.6 kPa excluded USS abnormality (98% negative predictive value). All patients with PH on USS had a TE of $\geqslant$10.8 kPa. Furthermore, USS were normal in 3 with oesophageal varices: all had raised TE.

Conclusion: TE can be used to identify CF patients likely to have changes of CFLD on USS, and may be a better predictor of significant disease than USS alone. Since it is quick to administer, gives immediate results and can be measured during routine outpatient review, TE may be a useful tool to guide which patients are referred on for more resource intensive investigations that require specialist staff to administer and interpret.

187 Evaluation of cystic fibrosis liver disease and the relation with risk factors in a Romanian centre

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Introduction: Liver disease is the second non-pulmonary cause of death in cystic fibrosis, witch, with increasing life expectancy, became an important management problem. Predisposing factors like male sex, pancreatic insufficiency, meconium ileus and severe mutation are incriminated to influence the occurrence of cystic fibrosis associated liver disease (CFLD).

Objectives: Evaluation of CFLD presence among CF patients and identification of its risk factors in Romanian population.

Methods: Study included 174 patients with CF, monitored in the National CF Centre. They were routinely followed-up by clinical assessment, liver biochemical tests, ultrasound examinations and other methods like transient elastography, biopsy, in selected cases. CFLD was diagnosed according current guidelines. A chi square test of independence with Yates continuity correction was used to determine the association between the risk factors and the presence of CFLD.

Results: Sixty-six patients, median age at diagnosis 4.33 years, were diagnosed with CFLD, without significant gender gap. CFLD was frequent in patients aged over 8 years, with meconium ileus history, carriers of severe mutations ($p = 0.002$). Pancreatic insufficiency, although present in 75% of patients with CFLD was not confirmed as risk factor, nor male gender, in our study.

Conclusion: CF children older than eight years of age, carriers of a severe genotype, with a positive history of meconium ileus, were more likely predisposed to CFLD. Further studies should discover other significant influences in order to prevent the development of liver cirrhosis and prolong the life of cystic fibrosis children.